Summary

- While DNA contains material common to all humans, some portions are unique to each individual.
- DNA profiling is a way of establishing identity.
- It is used in a variety of ways, such as establishing proof of paternity or finding out whether twins are fraternal or identical.
- Critics point out that DNA profiles are vulnerable to contamination errors, and invasion of privacy.

Genes make up the blueprint for our bodies, governing factors such as growth, development and functioning. Almost every cell in the human body contains a copy of the blueprint, stored inside a special sac called the nucleus. The estimated 23,000 genes are beaded along tightly bundled strands of a chemical substance called deoxyribonucleic acid (DNA). These strands are known as chromosomes. Humans have 46 paired chromosomes (half inherited from each parent), with two sex chromosomes that decide gender and 44 chromosomes that dictate other factors. Certain portions of DNA are unique to each individual. DNA profiling is a way of establishing identity and is used in a variety of ways, such as finding out whether twins are fraternal or identical. DNA samples are usually obtained from blood.

Uses of DNA profiling

Some of the uses of DNA profiling include:

- paternity - to find out if the alleged father is actually the biological father of the child
- twins - identical twins share the same genetic material, while fraternal (non identical) twins develop from two eggs fertilised by two sperm and are no more alike than individual siblings born at different times. It can be difficult to tell at birth whether twins are identical or fraternal
- siblings - for example, adopted people may want to have DNA tests to make sure that alleged biological siblings are actually their blood brothers or sisters
- immigration - some visa applications may depend on proof of relatedness
- criminal justice - DNA testing can help solve crimes by comparing the DNA profiles of suspects to offender samples. Victorian law allows the collection of blood and saliva samples from convicted criminals and suspects. DNA profiles are then kept on a database.

DNA profile explained

While DNA contains material common to all humans, some portions are unique to each individual. These portions, or regions, contain two genetic types (alleles) that are inherited from the person’s mother and father. A person’s DNA profile is made by investigating a number of these regions. In a paternity test, for example, the mother’s DNA profile is compared with the child’s to find which half was passed on by the mother. The other half of the child’s DNA is then compared with the alleged father’s DNA profile. If they don’t match, the ‘father’ is excluded, which means he isn’t the father of that child. If the DNA profiles match, the ‘father’ is not excluded - which means there is a high probability (more than 99 per cent) that he is the father. DNA tests such as this can’t offer 100 per cent proof.

Advantages of DNA profiling

Some of the advantages of DNA profiling include:

- DNA tests can be applied to any human sample that contains cells with nuclei, such as saliva, semen, urine and hair.
- DNA tests are extremely sensitive, and can be conducted using samples that would be too small for other serological tests.
- DNA is hardy, and resists degeneration even after contamination with chemicals or bacteria.
- The ability of DNA profiling to exclude a suspect means the police are able to confidently drop that line of enquiry and continue their investigation down other avenues.

Limitations of DNA profiling

Contrary to public belief, DNA profiling isn’t infallible. Critics point out various problems and limitations, including:

- New DNA profiling technologies can give incorrect results, due to errors such as cross-contamination of samples.
- Older DNA profiling technologies are more prone to errors, which could give false-negative or false-positive results.
- DNA profiles can only offer statistical probability (for example, one in a million), rather than absolute certainty.
- The more people tested, the lower the statistical probability. For example, the probability of one in a million may nosedive to one in 10,000 if enough people are profiled for a single test.
- DNA databases stored on computer are vulnerable to exploitation via hackers.
- Some critics point out that holding a person’s DNA profile on record is, in a sense, a violation of that person’s DNA ‘ownership’.
- DNA evidence is easily planted at a crime scene.

Where to get help

- Your doctor
- Victorian Clinical Genetics Services - Royal Children’s Hospital Tel. (03) 8341 6201
- Victorian Institute of Forensic Medicine Tel. (03) 9684 4444
Things to remember

- While DNA contains material common to all humans, some portions are unique to each individual.
- DNA profiling is a way of establishing identity.
- It is used in a variety of ways, such as establishing proof of paternity or finding out whether twins are fraternal or identical.
- Critics point out that DNA profiles are vulnerable to contamination errors, and invasion of privacy.

References

- DNA testing, Victorian Institute of Forensic Medicine Molecular Biology (VIFM), Department of Forensic Medicine (Monash University) & Department of Justice (State Government of Victoria), Melbourne, Australia. More information here.
- Paternity testing, Victorian Institute of Forensic Medicine Molecular Biology (VIFM), Department of Forensic Medicine (Monash University) & Department of Justice (State Government of Victoria), Melbourne, Australia. More information here.
- Action Now, Justice Action, NSW, Australia.

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Genes and genetics

The following content is displayed as Tabs. Once you have activated a link navigate to the end of the list to view its associated content. The activated link is defined as Active Tab

- A-Z of genetic conditions
- Genes and genetics explained
- Genetic testing

A-Z of genetic conditions

- Ambiguous genitalia
  The causes of ambiguous genitalia include genetic variations, hormonal imbalances and malformations of the fetal tissues that are supposed to evolve into genitals...
- Angelman syndrome
  The characteristic features of Angelman syndrome are not always obvious at birth, but develop during childhood...
- Ankylosing spondylitis
  Ankylosing spondylitis (AS) is a type of inflammatory arthritis that targets the joints of the spine...
- Barrett's oesophagus
  Symptoms of Barrett's oesophagus are similar to regular heartburn, which means many people don't seek treatment until their condition is quite advanced...
- Bipolar disorder
  Bipolar disorder is a type of psychosis, which means the person's perception of reality is altered. It is characterised by extreme mood swings...
- Central nervous system birth defects
  Folic acid taken before conception, and during at least the first four weeks of pregnancy, can prevent around seven out of 10 cases of neural tube defects...
- Charcot-Marie-Tooth disease (CMT)
  Charcot-Marie-Tooth disease is the most common inherited disorder affecting the peripheral nervous system...
- Cleft palate and cleft lip
  Most cleft palates and cleft lips can be repaired so that appearance and speech develop normally...
- Congenital adrenal hyperplasia (CAH)
  CAH is a rare genetic disorder, but it is well understood and treatment is readily available...
- Creutzfeldt-Jakob disease (CJD)
  Creutzfeldt-Jakob disease is characterised by physical deterioration of the brain, dementia and walking difficulties...
- Cri du chat syndrome
  Most children born with cri du chat syndrome have moderate intellectual disability, with varying degrees of speech delay and some health problems...
- Cystic fibrosis (CF)
When a person has cystic fibrosis, their mucus glands secrete very thick sticky mucus that clogs the tiny air passages in the lungs and traps bacteria.

- Digestive tract birth defects
  Too much amniotic fluid surrounding the baby during pregnancy (polyhydramnios) may indicate the presence of defects of the digestive tract.

- Down syndrome
  With the support and opportunities available to them today, most people with Down syndrome are able to achieve and participate as valued members of their community.

- Dwarfism
  Dwarfism refers to a group of conditions characterised by shorter than normal skeletal growth.

- Eczema (atopic dermatitis)
  Eczema can vary in severity, and symptoms may flare up or subside from day to day.

- Essential tremor
  Essential tremor causes involuntary shaking or trembling of particular parts of the body, usually the head and hands, but it is not Parkinson's disease.

- Fragile X syndrome
  The facts about fragile X syndrome are complicated, and parents and family members are invited to ask their doctor to refer them to a genetics clinic.

- Friedrich's ataxia
  To the casual observer, a person with Friedrich ataxia may seem to be drunk.

- Genetic factors and cholesterol
  Familial hypercholesterolaemia is an inherited condition characterised by higher than normal levels of blood cholesterol.

- Haemochromatosis
  Haemochromatosis (iron overload disorder) tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses.

- Haemophilia
  All children with severe haemophilia are given preventative treatment with infusions of blood products before they have a bleed.

- Hair
  Human hair grows one centimetre every month.

- Hearing problems in children
  The earlier that hearing loss is identified in children, the better for the child's language, learning and overall development.

- Huntington's disease
  The symptoms of Huntington's disease usually, but not always, first appear when the person is approaching middle age.

- Kabuki syndrome
  Kabuki syndrome affects males and females equally and there is no cure.

- Kennedy's disease
  Kennedy's disease is a rare inherited neuromuscular disorder that causes progressive weakening and wasting of the muscles, particularly the arms and legs.

- Kidney medullary cystic kidney disease
  Medullary cystic kidney disease causes the growth of abnormal cysts in the kidneys.

- Kidney polycystic kidney disease (PKD)
  Polycystic kidney disease is a common cause of kidney failure in Australia and equally affects men and women of different ethnic backgrounds.

- Klinefelter syndrome
  Klinefelter syndrome is often diagnosed at puberty, when the expected physical changes don't occur.

- Leukodystrophy
  Leukodystrophy refers to a group of inherited disorders that affect the white matter of the brain, which causes loss of normal brain functions.

- Long QT syndrome
  You should be investigated for long QT syndrome if you faint for no apparent reason, during or after exercise or emotional excitement.
- Marfan syndrome
  Some people may not realise they have Marfan syndrome, because their features are either mild or not obvious.

- McCune-Albright syndrome
  The severity of symptoms or how a child with McCune-Albright syndrome will be affected throughout life is difficult to predict.

- Muscular dystrophy
  People affected by muscular dystrophy have different degrees of independence, mobility and carer needs.

- Neurofibromatosis
  Neurofibromatosis is caused by faulty genes, which may be inherited or have spontaneously mutated at conception.

- Noonan syndrome
  Noonan syndrome is a genetic condition that usually includes heart abnormalities and characteristic facial features.

- Osteoporosis in children
  Osteoporosis in children is rare and usually caused by an underlying medical condition.

- Phenylketonuria (PKU)
  PKU is an inherited disorder that prevents the normal breakdown of a protein found in some foods.

- Porphyrin
  Porphyrin can affect the skin, nervous system, gastrointestinal system or all of these, depending on the specific type.

- Prader-Willi syndrome
  A feature of Prader-Willi syndrome is the child's excessive appetite, which often leads to obesity.

- Premature and early menopause
  The symptoms of premature or early menopause are the same as for menopause at any age.

- Rett syndrome
  People with Rett syndrome have a keen desire to communicate.

- Spinal muscular atrophy (SMA)
  A child with spinal muscular atrophy type 1 rarely lives beyond three years of age.

- Tay-Sachs disease
  Tay-Sachs disease is a serious genetic disorder common in Ashkenazi Jews and French-Canadians.

- Thalassaemia
  Thalassaemia is an inherited blood disorder that can cause anaemia or death if not treated.

- Tourette syndrome
  Milder forms of Tourette syndrome can be misdiagnosed, as it often occurs at the same time as attention deficit hyperactivity disorder (ADHD) and other disorders.

- Treacher Collins syndrome
  Treacher Collins syndrome is a genetic disorder that affects growth and development of the head, causing facial defects and hearing loss.

- Trisomy disorders
  Children affected by trisomy usually have a range of birth defects, including delayed development and intellectual disabilities.

- Tuberous sclerosis
  Tuberous sclerosis is a genetic disorder that affects various parts of the body to varying degrees of severity.

- Turner's syndrome
  Turner's syndrome is a random genetic disorder that affects females, causing short stature and infertility.

- Usher syndrome
  Services aim to help a person with Usher syndrome prepare for the dual loss of sight and hearing.

- Von Willebrand disease
A person with von Willebrand disease may have frequent nosebleeds, heavy menstruation or excessive bleeding from the mouth.

- **Williams syndrome**
  Williams syndrome often goes undiagnosed, which means that some people with the disorder fail to get the support and treatment they need until later in life.

- **Wilson disease**
  In Wilson's disease, a build-up of copper damages organs including the liver, nervous system, brain, kidneys and eyes.

**Genes and genetics explained**

- **Genes and genetics explained**
  Children inherit physical characteristics such as eye colour from their parents through their genes.

- **Gene therapy**
  Gene therapy is an experimental form of treatment that targets the faulty genes that cause genetic diseases.

- **Genetic disorders**
  Genetic disorder is caused by an altered or faulty gene or set of genes.

- **Genetic services in Victoria**
  Genetic services can help people who are affected by, or who are at risk of, inherited conditions or birth defects, to make informed choices about their healthcare.

**Genetic testing**

- **DNA profiling**
  DNA profiling is a way of establishing identity and is used in a variety of ways.

- **Egg freezing**
  You can freeze your eggs for medical reasons or for reasons that are more to do with your life circumstances.

- **Genetic services in Victoria**
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- **Genetic testing for inherited cancer**
  A predisposition to certain cancers can be inherited via altered genes.

- **Newborn bloodspot screening**
  Every newborn baby in Australia is offered a newborn bloodspot screening test to identify those at risk of rare, but serious, medical conditions.

- **Pregnancy tests – chorionic villus sampling**
  Chorionic villus sampling (CVS) is a pregnancy test that checks the baby for some abnormalities.

**Related Information**

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  Genetic services can help people who are affected by, or who are at risk of, inherited conditions or birth defects, to make informed choices about their healthcare.

- **Gene therapy**
  Gene therapy is an experimental form of treatment that targets the faulty genes that cause genetic diseases.

- **Pregnancy tests - maternal serum screening**
  Maternal serum screening can indicate increased risk of abnormalities in the unborn child, but is not a diagnosis.

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